FACT SHEET
Healthcare Provider

Biotinidase Deficiency

Description:
Biotinidase deficiency is an autosomal recessive metabolic disorder. Biotinidase limits the liberation and recycling of the vitamin biotin. Deficiency of the enzyme biotinidase results in improper functioning of carboxylase enzymes essential to the body's ability to alter fats and to metabolize carbohydrates and protein is impaired.

Incidence in General Population:
1:75,000 live births

Symptoms:
Severity
- Physical disabilities: seizures, skin rash, skin infection, alopecia, hypotonis, hearing loss, conjunctivitis, ataxia, breathing problem.
- Developmental disabilities: mental retardation, developmental delay.

Mortality
- Metabolic acidosis and organic acidemia can cause coma and death.

Variants
- Partial biotinidase deficiency is a milder form where activity of biotinidase is about 10-30% of the enzyme's normal activity. Symptoms usually do not develop except under periods of stress from infection or poor diet.
- Symptomatic diagnosis is difficult because the age of onset of symptoms may be anywhere between 1 week and 10 years of age. Usually symptoms appear between 3 and 5 months of age.

Diagnosis:
Newborn screening—A qualitative colorimetric assay is used to determine the presence of the enzyme. In the presence of the enzyme, a color change occurs. A second dried-blood-spot filter paper card may be requested by the Newborn Screening Laboratory if the initial screening result is above the normal range. Infants with presumptive positive screening (critical) results require prompt follow up. If this occurred, the clinician would be contacted by the Metabolic Treatment Center. When notified of these results, the clinician should immediately check on the clinical status of the baby and facilitate referral to the Metabolic Treatment Center. The Metabolic Treatment Center will provide consultation and assistance with diagnostic testing.

Monitoring:
Clinical observation is important for healthcare providers caring for patients with Biotinidase. It is important for primary care provider and the Metabolic Center to develop an ongoing collaborative relationship in caring for these patients.

Treatment:
The acute symptoms of biotinidase deficiency will disappear with administration of pharmacological doses of oral biotin, usually between 5mg and 20mg per day. This provides the body with sufficient free biotin for all metabolic needs. Therapy is lifelong, and no dietary restrictions are necessary. Prognosis is good for children diagnosed with biotinidase deficiency prior to the occurrence of symptoms. No serious side effects of biotin treatment have been recognized.

Illness and Immunizations:
Immunizations should be kept current. Consult with the Metabolic Center within 24 hours of the onset of an illness or at the time of hospitalization.

Growth and Development:
Monitor child for normal growth and developmental milestones.

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